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# Gaucher's Disease:

## A Nurse's Handbook

Clinical Center

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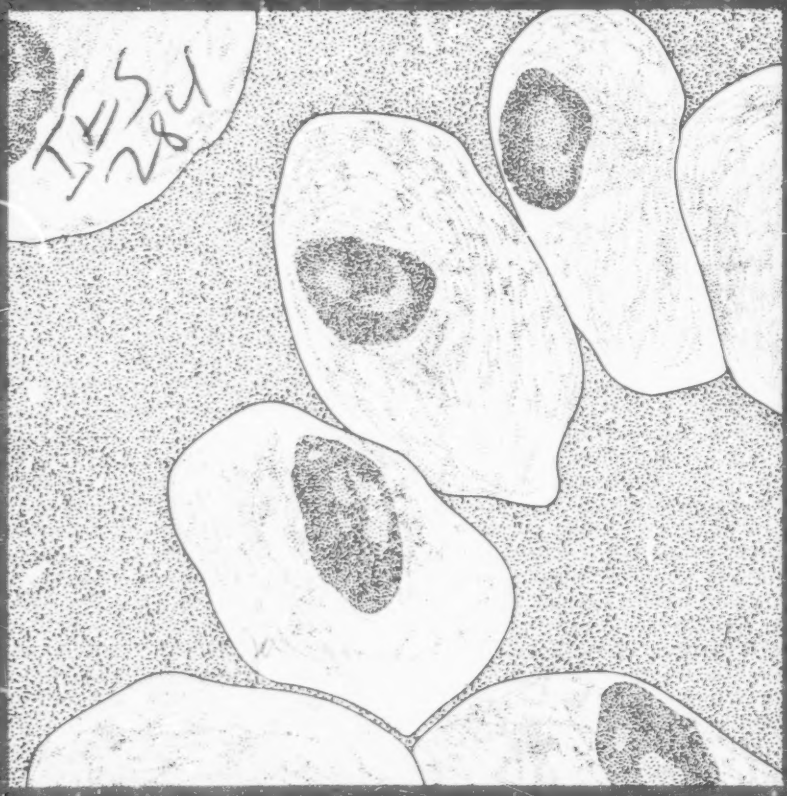

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## Handbook Objectives

1. Provide basic information regarding Gaucher's disease to the nursing staff.
2. Provide information to make assessments and nursing care more applicable to the needs of this patient population.
3. Improve the quality of the patient education programs for Gaucher patients by expanding the nurse's knowledge base of the needs of the Gaucher patient.

## Reader Objectives

1. State three symptoms characteristic of type 1 Gaucher's disease.
2. State the major similarities between type 2 and type 3 Gaucher's disease.
3. Describe three effects on, or changes in, a patient's lifestyle that may occur as a result of Gaucher's disease.
4. State at least three questions to ask during a patient assessment that would be relevant to an individual with Gaucher's disease type 1.
5. Devise a teaching plan that encompasses at least three educational needs that are relevant to this patient population.

## Introduction

This handbook provides basic information about Gaucher's disease: its history, causative factors, means of diagnosis, and description of the three types of the disorder. Management of symptoms will also be addressed. Suggestions for the nurse in terms of patient assessment will be presented as well as sample nursing care plans. This patient population is frequently in need of both physical and emotional support in the community so a list of helpful agencies is also included.

One of the major problems encountered by individuals diagnosed with Gaucher's disease is receiving accurate information about the disease. Nursing staff on the Neurology Nursing Service at the National Institutes of Health are in the unique position to educate these patients and assist them in maintaining their highest level of health. Although there are still many unanswered questions about Gaucher's disease, this handbook will provide basic information regarding the disorder. Your knowledge can then be used to complete a comprehensive nursing assessment and a nursing care plan which will facilitate patient and family teaching.

The handbook provides a history of the disorder, information on the cause and types of disease, means to diagnose it, and signs and symptoms presented in patients. Questions raised by patients as well as treatment methods are also addressed. Nursing care of the patient is provided in terms of assessment and care planning. A glossary is also included.

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## History

Phillipe Gaucher first recognized this disease in a patient with hepatosplenomegaly in 1882. Other patients with similar problems continued to be reported and, in 1900, familial incidence of the disease was documented. In 1924, researchers began to investigate stored fatty material in body organs. A. Aghion documented increased amounts of glucocerebroside, a specific sphingolipid, in the body tissues of these patients. In 1965, Brady and associates documented the cause of the disorder as a deficiency of the lysosomal enzyme, glucocerebrosidase (Brady, 1965). The gene for this enzyme has been isolated and is being used in a variety of diagnostic and treatment protocols (Ginns, 1984) Many research centers throughout the world continue to study the mysteries surrounding the disease.

## Cause and Types

Lysosomes are intracellular organelles. One of their functions is the breakdown of cellular materials. They are present in all body cells but are particularly abundant in the reticuloendothelial system, which plays a primary role in the process of phagocytosis. The lysosomes use enzymes to degrade cellular components (Hers and Van Hoof, 1973).

In Gaucher's disease, the lack of the enzyme glucocerebrosidase causes a reduction in the breakdown of glucocerebroside. This material accumulates in the lysosomes. Glucocerebroside results from the normal turnover of cells in any tissue (Brady and Barranger, 1983). White and red blood cells are probably the main source of the accumulating lipid (Brady and Barranger, 1983).

Gaucher cells are very characteristic in appearance and their presence strongly suggest the disorder. These cells contain lipid and have cytoplasm with a "wrinkled tissue paper" appearance. The cells are in all parts of the reticuloendothelial system and arise from monocyte precursors in the bone marrow (Brady and Barranger, 1983). Cells that are similar in appearance may be found in patients with other disorders such as multiple myeloma, leukemia, thallassemia, and congenital dyserythropoietic anemia (Brady and Barranger, 1983). Diagnosis of the disease is confirmed by the deficiency of the enzyme activity. Assays are performed conveniently on white blood cells or cells grown from a skin biopsy.

The patients with this metabolic problem are grouped into three types. Type 1, the chronic non-neuronopathic form, was previously known as the "adult" form of the disease. It is the most common form and is more frequent in the Ashkenazic Jewish population. Carriers of the gene in this population may be as frequent as 1 in 12. Signs and symptoms may occur at any time after birth. The major signs of this form of Gaucher's disease are: splenomegaly, leukopenia, anemia, and thrombocytopenia related to hypersplenism, hepatomegaly, and bony changes with bone pain. Most of these patients live a normal lifespan.

Type 2, the acute neuronopathic form, was previously known as the

"infantile" form of the disease. It is usually recognized at several months of age in an infant who appears normal at birth and thrived initially. It is seen in the same numbers in all populations and is rare in occurrence when compared to type I disease. The presenting signs are hepatosplenomegaly, a rigid or hypertonic status of the muscles, strabismus, trismus, retroflexion of the head, respiratory instability, and sometimes seizures. Death occurs by the age of 2 years and usually is a result of respiratory failure such as apnea or aspiration. Bone changes are rare. Gaucher cells are found in the perivascular area (Virchow-Robin spaces) of the central nervous system and within the substance of the brain. Neurons characteristically exhibit diffuse degenerative changes, but gliosis is mild in comparison to other storage diseases.

Type 3, the subacute neuronopathic form, was previously known as the "juvenile" form and also is very rare. This type shares characteristics of type 1 and type 2, but has a more prolonged course. It has not been well described clinically (Nishimura, 1980). Complications involving the central nervous system may include: seizures, myoclonus, oculomotor apraxias, ataxia, spasticity, and mental retardation (Nishimura, 1980).

It has been shown that the various types of Gaucher's disease are caused by different alterations in the structure of the glucocerebrosidase molecule (Ginns, 1982).

## Diagnosis

The diagnosis of Gaucher's disease involves the evaluation of several patient parameters.

- Splenomegaly, and usually hepatomegaly, are present in almost all cases.
- Bone marrow aspirates will show Gaucher cells (Numbers of the cells do not correlate with severity of the disease.)
- An elevation of serum nontartrate-inhibitable acid phosphatase in the blood stream is frequently seen.
- Glucocerebrosidase activity can be directly measured by using leukocytes in venous blood or by using cultured skin fibroblasts.
- Glucocerebroside in the tissues can be directly measured via liver biopsy. The tissue levels of the lipid do not correlate with clinical presentation.

Carriers of the disorder may also be diagnosed by use of the blood test or skin biopsy to measure glucocerebrosidase activity. Prenatally, the activity can be measured by culturing amniocytes via amniocentesis (Brady and Barranger, 1983).

Recent research at the National Institutes of Health has revealed means to detect whether or not a patient will develop neurologic symptoms (Ginns, 1983). These tests could lead to more accurate prenatal diagnosis and predictions about complications.

Patients may go through numerous diagnostic studies to confirm the diagnosis and gauge the extent and rate of progression of their disease. These studies may include some of the following:

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**Table I**
**Diagnostic Studies**

| Test | Rationale |
|---|---|
| Bone marrow aspiration and biopsy | To document presence of the Gaucher cells in the bone marrow/bone |
| Liver biopsy | To document presence of the Gaucher cells in the liver tissue, detect liver changes, to measure glucocerebroside in the liver |
| Bone x-rays | To assess the degree of structural changes in bone related to the disease |
| Liver-spleen scan | To measure the exact size of the liver/spleen, to assist in monitoring of changes in size of the organs |
| Abdominal ultrasound | To examine the liver and spleen |
| Electroencephalogram | To document seizure activity |
| Eye examinations | To observe for pingueculae and abnormal eye movements |
| Skin biopsy | To directly measure glucocerebrosidase activity in cultured skin fibroblasts |
| Blood tests—CBC, SMAC, coagulation studies, immunoglobulin level, glucocerebrosidase levels | To assess hematologic status, liver function, immune status, and to diagnose the disorder by determining enzyme levels using leukocytes |
| Bone scan | To assess structural change and abnormalities in the skeleton |

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## Signs and Symptoms

### Type 1

It is important to stress in the beginning of this section the wide range of symptoms that may be present in any patient. Some patients have few symptoms, others many. Some people, for example, may have mainly bone problems, others have only an enlarged spleen. Symptoms can be present in any combination, and it is important to remember the individuality of each patient's care. The following is a brief description of all symptoms or signs that may occur. Please remember that each patient may not exhibit all these symptoms (Sack, 1980). The signs/symptoms will be grouped according to the 13 assessment categories used at the Clinical Center by the nursing staff.

### Type 1

*Air*—Gaucher cells can be found in the lungs of affected individuals. However, most patients do not have a greater incidence of respiratory infection and pneumonia. Severe pulmonary problems are very rare but hypoxia, cyanotic appearance and clubbing of the fingers may occur in these patients.

*Circulation*—One of the most common symptoms is splenomegaly. The enlargement is usually painless, and the spleen usually contains fibrous tissure. Rupture of the spleen is not a common occurrence.

Changes in blood counts may occur secondary to changes in the spleen. Thrombocytopenia, anemia, and leukopenia may be present. Frequent infection may occur. Bleeding problems secondary to the changes in platelet numbers may require blood and/or platelet transfusions for some patients. Splenectomy usually resolves the problems of pancytopenia.

Many individuals with Gaucher's disease also report other symptoms related to their circulation. Children may have frequent episodes of epistaxis. These seem to become less of a problem as the children mature. Individuals also report that they frequently get bruises, especially on the lower extremeties. The episodes of epistaxis and ecchymosis are for the most part the result of the thrombocytopenia, but they may occur in patients with a normal platelet count.

Hepatomegaly is also a common finding. Liver function tests may also be abnormal. It has been reported that the amount of hepatic involvement correlates with the overall severity of the disease. Cirrhosis, esophageal varices and gastrointestinal hemorrhage have been reported (James, 1981). Disorders of the immune system may be present and cause increased susceptibility to infection. Although uncommon, several tumors have also been associated with Gaucher's disease: multiple myeloma, leukemia, astrocytoma, lung tumors, and Hodgkin's disease.

*Food/Fluid*—Gaucher patients may have gastrointestinal complaints. Children frequently have poor appetites. Fatty meals may be difficult to digest.

*Elimination*—Diarrhea may be a problem, especially after large or fatty meals.

*Neuro/Sensory*—Type 1 patients have a normal neurological examination.

*Sleep/Rest*—Type 1 patients may have sleep difficulty related to bone pain or emotional problems.

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*Physical safety*—Patients who are thrombocytopenic will be predisposed to hemorrhage if physical injuries occur. Patients with severe bone disease also will be predisposed to pathological fractures from trauma. Patients with gait disturbances due to skeletal changes also will be at risk for injury from falls.

*Mobility*—Changes in the skeleton are a common finding in patients with type 1 disease. Bones appear eroded on x-ray films and many deformities are apparent. Pain and decreased range of motion may also occur as a result of aseptic necrosis of the femoral head.

Pathologic fractures of bone and spinal deformities from changes in the vertebral column are observed. Many patients have a deformity of the lower femur, known as the Erlenmeyer flask deformity, which is observed as an expansion of the femur on knee x-rays (Goldblatt, 1978).

*Comfort/Pain*—Bony problems are the most likely to produce discomfort and disability. Dull aches in the bone, as well as joint pain can occur. Some patients experience episodes of severe bone pain that often are mistaken for osteomyelitis. The pain is centered in one area of the bone which is frequently warm, red, and swollen. Patients may be pyrexic. The pain may last for as long as 2 weeks. The attacks are episodic in nature and usually self-limited (Goidblatt, 1978).

*Hygiene/Skin*—The skin of some patients may have a yellowish brown coloring. The color may be more intense in the lower extremities. The cause of the increased pigmentation is not known. The patients may note increased tanning during the summer. Yellowish deposits on the sclera of the eye called pingueculae are frequently observed. They are small fatty deposits on the eye that are not harmful, in fact, many people who do not have Gaucher's disease have pingueculae.

Dental problems may also occur. Children seem to keep their baby teeth for a longer period of time and may have delayed appearance of their permanent teeth.

*Sexuality*—(Several observations are related to maturation). Children with Gaucher's disease may be smaller in stature than others in the same age bracket. Provided they are not otherwise ill, the eventual height of Gaucher children is expected to be normal. Puberty may also be delayed, creating late onset of menstrual periods and slow appearance of secondary sex characteristics.

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**Type 2**
As stated previously, the signs and symptoms of type 2 disease are usually present by 3 months of age.

*Air*—Due to neurologic deficits, the infants may exhibit periods of apnea and require suctioning to handle secretions that they have difficulty swallowing. They are predisposed to aspiration because of the decreased gag reflex. Many of the infants die of respiratory infection or apnea.

*Circulation*—Hepatosplenomegaly is present.

*Food/Fluid*—Due to neurological deficits, the infants frequently have a poor sucking reflex and have difficulty with swallowing. They may require feeding via nasogastric tube or gastrostomy tube.

*Neuro/Sensory*—These infants have neurologic involvement. Signs may include: trismus, retroflexion of the head, seizures, strabismus, hypertonicity, pathologic reflexes, and apathy (Brady and Barranger, 1983).

*Mobility*—Skeletal changes are not usually present. The infants will have difficulty performing the normal physical milestones for their age due to nervous system involvement (Lifting head, sitting, turning over).

**Type 3**
The signs and symptoms of type 1 disease are also characteristic of type 3 but additional symptoms include:

*Neuro/Sensory*—Neurologic symptoms occur but are not as severe as type 2 disease and can occur from the first to the fourth decade in life. The course is more chronic than type 2 disease. Common neurologic symptoms include: supranuclear ophthalmoplegia, seizures, myoclonus, mental retardation, spasticity, pathologic reflexes, strabismus, ptosis, dysphagia, and dysphasia (Nishimura, 1980). Nervous system involvement may create difficulties with feeding, mobility, communication, activities of daily living, and respiratory status. Each patient will have individual problems, all based on the area of the nervous system that is affected.

**Table 2**
**Signs and Symptoms of Gaucher's Disease**

**Type 1**

*Air*—Occasionally increased incidence of respiratory infection
Severe - hypoxia, cyanosis, clubbed fingers/toes.

*Circulation*—Splenomegaly
Thrombocytopenia, leukopenia, anemia.
Epistaxis in children
Bruises on lower extremity
even with normal platelets
Hepatomegaly
Severe cirrhosis, esophageal varices, GI bleeding.
Immune system - increased infection and association with several tumors.

*Food/Fluid*—Poor appetite with children. Fatty foods cause GI complaints.

*Elimination*—Diarrhea after large or fatty meals.

**Neuro/Sensory*— Normal neurological exam

*Mobility— Bony erosion. Aseptic* necrosis of femoral heads. Pathologic fractures. Spinal deformities due to vertebrae column changes. Erlenmeyer flask deformity.

*Comfort/Pain*—Dull aches

Severe bone crisis (episodic)

*Hygiene/Skin*—Increased Pigmentation Pingueculae

*Sexuality*— Children—Short stature delayed appearance of permanent teeth delayed puberty/menarche.

**Type 2**

*Air*—Periods of apnea, dysphagia, predisposed to aspiration. Often die of respiratory infection.

*Circulation*—Hepatosplenomegaly

*Food/Fluid*—Poor suck, dysphagia, may need NG feedings/gastrostomy.

**Neuro/Sensory*—Trismus, retroflexion of head, seizures, strabismus, pathologic reflexes, hypertonicity, and apathy.

*Mobility*— Skeletal changes are usually not present.

**Type 3**

Same as Type 1 disease

**Neuro/Sensory*—Between first and fourth decade of life: supranuclear ophthalmoplegia, seizures, myoclonus, mental retardation, spasticity, pathologic reflexes, strabismus, ptosis, dysphagia, and dysphasia. The nervous system involvement may affect mobility, communication, feeding, activities of daily living, and respiratory status.

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### Issues Raised by the Patient

There are several topics that may be of interest to this patient population. Splenectomy is a topic that frequently arises in the course of a patient's hospital stay. Many have been informed that splenectomy causes increased hepatic and bone disease since the storage material seeks other areas of deposit. There is much controversy about whether splenectomy causes progression of the disease. The conservative approach is to keep the spleen as long as possible. One of the indications for splenectomy is bleeding problems such as frequent epistaxis, gum bleeding, hematuria, melena, and gastrointestinal bleeding. Splenomegaly may also create pressure on the lungs and cause dyspnea. Pressure on the gut and large blood vessels in the abdomen may also occur. More severely affected children may have anemia and cardiac failure. The need for splenectomy is decided by the physician after carefully weighing all signs and symptoms. Some patients may view splenectomy as a "life and death" situation. They will need explanations about the rationale for splenectomy and how the procedure can be expected to affect their health and their disease process.

One of the most frequently asked questions by patients is how the disease will affect their lifespan. In most cases of type 1, the disease is compatible with a normal life-span. In fact, many cases probably never come to medical attention. However, many cases of a more severe variety are known. Survival then depends on the complications. Type 2 is a severe neurologic disease with death occurring by the age of 2 years. In type 3, survival is more variable.

As stated previously, this disorder is inherited. Many questions arise regarding heredity and concern for offspring. The following diagrams will illustrate various possibilities in relationship to the disorder.

gg - individual with Gaucher's disease
Gg - carrier of the disease
GG - normal individual

| | g | g |
|---|---|---|
| G | Gg | Gg |
| g | gg | gg |

The example demonstrates how the various genetic possibilities are graphed by use of the Punnett square.

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| | G | g |
|---|---|---|
| G | GG | Gg |
| g | Gg | gg |

If two carriers marry, there is a 25 percent chance of a baby with the disease, 50 percent chance of a baby who is a carrier, and 25 percent chance of a normal baby.

| | g | g |
|---|---|---|
| G | Gg | Gg |
| g | gg | gg |

If an individual with the disease marries a carrier: there is a 50 percent chance of a baby with the disease, and a 5υ percent chance of a baby who is a carrier.

| | g | g |
|---|---|---|
| g | gg | gg |
| g | gg | gg |

If two individuals with the disease marry, all children will have the disease.

| | G | g |
|---|---|---|
| G | GG | Gg |
| G | GG | Gg |

If a carrier and a normal individual marry, there is a 50 percent chance of a baby who is a carrier, and a 50 percent chance of a normal baby.

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Since the disease is inherited, the diagnosis can create numerous concerns. Many parents are totally unaware of the fact that they are carriers. Families, especially the parents of a child with Gaucher's disease, may experience guilt feelings about passing on the disorder. Some parents may have difficulty accepting that they are carriers or that they have offspring with the disease. Many emotions can be elicited when it is learned that a diagnosis of Gaucher's disease has been made in the family. Family members may be concerned that they also are carriers or have the disorder. It can be a stressful time for the entire family. Family members will also need to be aware that genetic counseling is available.

Many patients with Gaucher's disease must learn to deal with changes in their body image. Children frequently must face being shorter than their peers. If dental problems occur, these children may need to be fitted for braces. Also, delays in puberty and menstrual periods can be stressful for adolescents. Many patients of all ages must deal with having an enlarged abdomen secondary to hepatosplenomegaly. Skin may be pale due to anemia or pigment changes can cause dark blotches on the complexion. Surgery for splenectomy or hip replacement creates scars and the loss of body parts. Deformities from fractures, changes in gait, and the need for ambulatory aids can also be difficult adjustments to make. These psychological adjustments can be as important to the patient as the actual physical changes that occur.

It is apparent that there are many emotional impacts on the patient and family. The discovery of an abnormality, completing the diagnostic testing, coping with the pain and other body changes, facing the possibility of surgery, and assuming the responsibilities associated with an inherited illness can all be great stresses. Gaucher's disease is also a chronic illness, which means the disorder is one that must be faced throughout one's life. These emotional stresses are certainly as important as the physical effects of the disease. They demand as much, and at times, even more attention from health care providers, patient, and family.

### Treatment Methods

At the present time, there is no cure for Gaucher's disease. Treatment is directed toward the prevailing symptoms. With the wide range of symptoms and severity encountered with this disease, intervention for each patient will be very different.

Patients with Gaucher's disease will need to have spleen and liver size monitored. Observations should also be made for problems with bleeding, infection, anemia, and liver function. Patients should be aware to observe for the signs and symptoms and to report any to their physician. Patients should also be informed to protect against traumatic injury to the abdomen. The spleens of Gaucher's patients are usually somewhat hard and do not easily rupture, but with increased spleen size it is sensible to try to avoid sharp blows to the midsection. Problems with repeated episodes of epistaxis may need referral for cauterization.

Patients may require splenectomy for reasons previously stated. A dramatic rise in blood counts usually occurs after splenectomy. Due to the increased risk of pneumococcal infections after splenectomy, patients should receive one injection of Pneumovax® preoperatively. This should not be repeated because of the significant local reactions.

Patients with liver disease will also need to be monitored for complications. Liver size, liver enzymes, and signs of gastrointestinal bleeding from esophageal varices should be monitored.

Many patients will have to deal with problems related to the bony complications. Management of pain may be one of the most important issues. Often the pain is minor and can be relieved simply by resting the painful area. But pain can be more chronic in nature and require modifications in activity and medication. Patients may need to alter exercise programs and participate in sports, such as swimming, walking, and bicycle riding that prevent stress to joints and bones.

Bony crises are also problems for some patients. The pain can be very severe; treatment includes analgesia and rest of the affected limb. If the pain is in the back, hip, or leg, bedrest may be indicated. Pain in the arm may require the use of a sling to support the limb. Tylenol® and fluids can be helpful for fever. It may take several weeks for the affected area to return to a "normal" state after the initial pain has subsided. For example, the patient may note a limp or decreased joint motion in the arm even after the pain has stopped. It is very stressful to have to deal with the possibility of these episodes occurring usually without warning. The episodes may continually involve one area, or could involve different areas. It is important to stress that the pain is self-limiting and will diminish.

Fractures due to weakened bones require intervention. Splints and casts are usually used after the bone is set. It is important to follow the instructions of the orthopedist since Gaucher bone takes longer to heal and is more predisposed to infection. Vertebral collapse can be a medical and surgical emergency. Failure to respond to this occurrence with appropriate acute care could result in permanent neurologic deficits.

In some cases, bone changes, may require the use of special aids to assist with mobility. Supports, such as braces, canes, crutches, and wheelchairs may be needed. Patients confined to wheelchairs will also need instruction on how to prevent decubiti.

Surgery in Gaucher patients is usually a last resort but clearly does hold a place in management of bony complications. Hip replacements are considered and should be done after careful consideration of risk of infection and delayed healing, pain, decreased function of the limb, life style, and bone condition.

The delayed appearance of puberty and short stature can be distressing problems for children. Sometimes it is helpful for them to meet others with the disorder--they can see for themselves that growth and development is not necessarily retarded. Hormonal tests can be done if there is a

question of an endocrine or glandular abnormality. But reassurance, support, and counseling are important parts of helping children and teenagers through these problems. Emotional support is also needed to help patients and families deal with body image changes, chronic illness, and changes in the family itself due to illness.

Accurate information regarding the genetics of the disorder also needs to be explained to the patient and family. Means for screening should also be provided so that families are aware of how to obtain diagnosis for themselves and future spouses.

In patients with type 2 and 3 disease, a neurologist would be consulted to deal with manifestations related to the central nervous system. Anticonvulsant therapy would be initiated for those with seizures. Other problems that may be encountered as a consequence of neurological disturbance may involve feeding, airway maintenance, and infection. Treatment is supportive in nature.

Organ and bone marrow transplantations are areas of treatment being explored. Research has also been done in the area of enzyme replacement. Gene therapy is being studied.

## Research

Basic research on Gaucher's disease has been initiated at various centers throughout the world. Research at the National Institutes of Health is conducted by the Developmental and Metabolic Neurology Branch of the National Institute of Neurological and Communicative Disorders and Stroke. Currently the Gaucher patients are involved in several research protocols. Patients may be admitted for observation and study of the disease. Numerous diagnostic and research tests are performed. Tests are done to confirm diagnosis and to gather information about the disorder. Research is also done on the use of intravenous enzyme replacement with glucocerebrosidase as a treatment modality. Plans for genetic replacement are being formulated.

### Nursing Care

Your initial assessment of a patient with Gaucher's disease should address symptoms and concerns of this specific patient population. The following is a list of reminders, by category, to keep in mind while interviewing the patient and family. These nursing assessment categories are routinely used for all patient assessments done at the National Institutes of Health, Clinical Center Nursing Department.

| Category | Reminders |
|---|---|
| **Air** | Signs of hypoxia, cyanotic skin/nails/lips circumoral pallor<br>Dyspnea<br>Changes in chest size and shape<br>Activity tolerance<br>Need for special positioning<br>Need for special respiratory care |
| **Circulation** | Fever/signs of infection<br>Signs of unusual bleeding<br>Signs of anemia<br>Edema in lower extremities if abdomen is enlarged<br>Ascites if liver involvement is severe<br>If splenectomized - vaccination with Pneumovax®<br>Assess CBC/liver enzymes/coagulation studies |
| **Food/Fluid** | Assess appetite<br>Ability to eat regular meals if abdomen is enlarged<br>Means for feeding if patient is an infant with dysphagia |
| **Elimination** | Assess hematuria/melena if thrombocytopenic<br>Assess stool frequency<br>Assess for constipation if on narcotics |
| **Neuro/Sensory** | Assess history of seizures<br>Observe for oculomotor disturbances<br>Assess pain, loss of sensation, motor difficulty, loss of bladder or bowel control, and abnormal reflexes in extremities |
| **Sleep/Rest** | Assess need for seizure precautions<br>Assess predisposition to pathologic fractures<br>Assess for dysphagia |
| **Mobility** | Assess for limited range of motion<br>Assess for gait disturbance<br>Assess use of aids/prosthesis<br>Assess for physical therapy regime |
| **Comfort/Pain** | Assess for abdominal or bone pain<br>History of bone crisis<br>Measures to aid pain |
| **Hygiene/Skin** | Observe for pigment changes<br>Observe for pingueculae<br>Assess need for assist with ADL<br>Assess for predisposition to decubiti |

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| | |
|---|---|
| **Sexuality** | Assess if patient has entered puberty<br>Assess if menses has begun<br>Assess awareness of genetic implication of the disease<br>Changes in sexual patterns secondary to pain |
| **Socio/Psych** | Problems related to body image<br>How is family dealing with illness<br>Is there an impending loss of an infant<br>Assess coping with chronic illness<br>Has the patient met others with the disorder<br>Awareness of organizations<br>Home tutoring |
| **Teaching/Learning** | Medications<br>Signs of hypoxia<br>Suctioning at home<br>Prevention of bleeding/infection<br>Dealing with fatigue<br>Decreasing edema in lower extremities<br>Monitor ascites at home<br>Need for Pneumovax® for splenectomized patients<br>Indications for splenectomy<br>Dietary consult for changes in eating behavior<br>Use of NG/gastrostomy tubes at home<br>Monitoring stool and urine for blood at home<br>Safety measures associated with seizures<br>Physical therapy consult to learn exercise routine<br>Occupational therapy consult to teach use of ADL aids<br>Purpose of hip replacement surgery<br>Nature of bone crisis<br>Proper use of analgesics<br>Means to decrease pain: thermal, relaxation, etc.<br>Genetic counseling and diagnosis<br>Information about delayed puberty<br>Teaching about diagnostic tests<br>Informing patient/family about organizations |

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Patients and their families are eager to hear about the results of the diagnostic studies. They also are in need of information about the disorder itself. Suggest that the patient write questions for the physician. The nurse can also provide information. Please remember not to give inaccurate information. Since many of the patients do not know others with the disease, it may be helpful to introduce them to other patients on the Service with Gaucher's disease.

Patients who are returning home may need referrals to their local agencies and may need equipment for home care. Discharge planning should be implemented as soon as possible.

Two sample assessments and care plans are included as examples.

## Summary

This handbook has presented basic information about Gaucher's disease and has highlighted areas of the nursing assessment that are pertinent to this patient population. This should assist with the development of relevant nursing care plans and the identification of teaching/learning needs for these patients. Since currently there is no cure for this disorder, nursing staff can provide continued support to this population of patients by assisting them to understand, cope, and manage their symptoms and emotional reactions to the disease.

**NAME:** Patient A Appendix 1

**DIAGNOSIS:** Gaucher's disease Type 1

**I. General Information**

**A. Admission vital signs: 37-90-26**

1. Temperature: 37
2. Pulse: 90 and regular
3. Respirations: 26 and regular
4. Blood Pressure: 130/80
5. Height: 5'5"
6. Weight: 140 lbs.

**B. Reason for admission: Gaucher diagnostic tests for screening.**

**C. Medications:**

Pencillin VK. 250mg 1 bid
Tylenol 325 mg, 2 tabs q 4-6 hours prn
Tylenol #3, one to two tabs prn

**D. Allergies: None**

**II. Assessment**

**A. Air**

1. Respiratory problems? States she has noted SOB on exertion
2. Cough? None, no signs of cyanosis
3. Aids? Does not walk long distances.

**B. Circulation**

1. Diagnosed Gaucher's as a child by bone marrow biopsy after series of infections. Had splenectomy at age 23 due to lowered blood counts - had no bleeding however. Her blood counts have lowered over the years - bruises easily on legs.
2. Swelling? Mild edema in lower extremities elevates them as much as possible.
3. Gets "a cold easily", no overt bleeding problems. States she has an enlarged liver - this has caused an increase in her abdominal girth. No jaundice, states that her "liver blood tests" are very abnormal.
4. Aids? Daily penicillin, elevates legs.

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**C. Food/Fluid**

1. Eating problems? Has difficulty eating a large meal, eats small amounts 4 times a day.
2. Problems with food preparation? She and husband both cook.
3. Appetite? Fair
4. Nausea/vomiting? None
5. Weight change? Stable over last year.
6. Special diet? Small frequent meals

**D. Elimination (Bowel/Bladder)**

1. *Bladder*
   a. Dysuria? None
   b. Incontinence/retention? None
   c. Burning? None
   d. Hematuria? None
2. *Bowel*
   a. Pattern (last stool?) qod
   b. Incontinence? None
   c. Blood in stool? None
   d. Regimen? If no stool - uses Metamucil or Senokot (takes tylenol #3)

**E. Neuro/Sensory**

1. Increased strength/weakness of any muscle group? 5/5 throughout, difficult to test R LE due to pain
2. Level of consciousness and orientation? A & O x 3
3. Verbal and written comprehension? No problems
4. Speech problems? None
5. Involuntary movements/rigidity? None
6. Type of seizures? None
7. Loss of sensation to heat, cold, touch or position? Intact
8. Visual changes? None, EOM's intact
9. Hearing changes? Normal
10. Taste changes? None
11. Family history of neurological disease? None

**F. Sleep/Rest**

1. Problems? Occasional problems due to pain
2. Aids? Uses analgesic at bedtime

**G. Physical Safety**

1. Problems? No falls, no fractures
2. Aids? Uses cane

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**H. Mobility**

1. Limitations? Has difficulty walking distances, has limp
2. Aids? Cane Limited in activities such as shopping as she does not have a wheelchair.

**I. Comfort/Pain**

1. Pain description? Dull ache in right hip, pain increases with ambulation
2. Pain precipitated by? Walking distances
3. Pain relieved by? tylenol #3, tylenol q4-6° prn, uses 3-4 tylenol #3/day

**J. Hygiene/Skin**

1. ADL management? Independent, has grab bar on her shower stall
2. Skin integrity? Intact, splenectomy scar
3. Oral mucosa? Intact
4. Ecchymotic areas? On Legs
5. Self-help devices? Grab bar in shower, rubber treads on shower floor

**K. Sexuality**

1. Any recent problems with sexual function? Ye· pain has affected her sexual relationship with husband, positions can be painful (hip pain)
2. Any need to discuss birth control? Have 2 children, has been through menopause
3. Genetic counseling? Aware of genetic component of the disease, both children are carriers and have had their spouses tested before marriage

**L. Socio/Psychological**

1. Emotional status? 45 years old, husband is supportive as are her children. Lives with husband in Toledo, Ohio. Has hematologist and orthopedic MD at home. Involved in P.T. program at a local facility. Finds her chronic pain very depressing, has been dealing with it for 7 years.
2. Are you seeking/receiving any counseling? No. Her local MD suggested therapy to her, she has not pursued this for financial reasons. States she has medical insurance.

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3. How are you managing with your illness? Has met two other patients with the disorder. She is Jewish. Her parents were upset when first diagnosed - we're not aware of the disease - overprotective to her as they thought illness was fatal - she feels here relationship with them is better. Works part-time as a secretary - has worked there 10 yrs., boss very understanding of her, has no problems in terms of job/pain, doesn't need to walk distances there, uses tyl. #3 if needed. Is able to drive.
4. What concerns you most about your disease? Will she need a hip replacement procedure? Would it benefit her?
5. Who is most supportive? Husband/close friends
6. How does your illness affect your occupation? No

**M. Teaching/Learning**

1. Knowledge and understanding of disease? Most concerned if her hip x-rays appear worse and if liver is larger
2. Knowledge and understanding of medications? Not aware that codeine can cause constipation
3. Knowledge and understanding of safety measures? Stated 3 sx's of infection and bleeding, follow-up with LMD on all fevers, understands why she needs penicillin to prevent pneumoccal infections (splenectomized).
4. What do you see as your most important learning need? Information regarding tests and results

**N. Other Medical Problems?**

None

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## NURSING CARE PLAN

Appendix 2

**REAL & POTENTIAL PROBLEMS:**

| | | | |
|---|---|---|---|
| **06/18** | **LACK OF KNOWLEDGE: R/T—RELATIONSHIP OF CONSTIPATION AND ANALGESIA.** | | |
| EXPECTED OUTCOMES | | C/P: | D/L: |
| 06/18 | —V-UNDERSTANDING OF RELATIONSHIP OF CONSTIPATION AND PAIN MEDS | T/TH | 6/23 |
| 06/18 | —V-BOWEL REGIME TO USE AT HOME. | T/TH | 6/23 |
| NURSING ACTIONS | | | |
| 06/18 | —DISCUSS THE EFFECT OF CODEINE ON THE INTESTINAL TRACT. | | |
| 06/18 | —DISCUSS PLANS FOR BOWEL REGIME AT HOME: EXPLORE USUAL TIME FOR BM'S, AIDS USED IN THE PAST, DISCUSS DIETARY MEANS TO AID CONSTIPATION (PRUNES, PRUNE JUICE, ROUGHAGE, FRESH FRUIT/VEG, BRAN, WHOLE GRAINS, ADEQUATE FLUIDS), USE OF WARM DRINKS, SETTING TIME ASIDE EACH DAY FOR BM. AIDS TO USE IF NO BM. | | |
| 06/18 | —HAVE PT. BEGIN STOOL RECORD SHE WILL KEEP AT BEDSIDE WHILE HOSPITALIZED. | | |
| 06/18 | **PAIN: R/T—ASEPTIC NECROSIS OF RIGHT HIP** | | |
| | EXPECTED OUTCOMES | C/P: | D/L: |
| 06/18 | IDENTIFIES & USES SEVERAL TECHNIQUES TO CONTROL PAIN | QD | 6/26 |
| 06/18 | —PT. WILL V-SEVERAL POSITIONS THAT MAY BE MORE COMFORTABLE FOR HER DURING INTERCOURSE. | T/TH | 6/23 |
| NURSING ACTIONS | | | |
| 06/18 | ASSESS TYPE, LOCATION, INTENSITY, & FREQUENCY OF PAIN | | |
| 06/18 | PAIN CONTROL PROGRAM—MEDICATE PT. WITH TYL #3 AT HS, PROVIDE MED PRN. | | |
| 06/18 | EXPLAIN NEED TO ASK FOR PRN MEDS | | |
| 06/18 | —TO USE CANE FOR AMBULATION, TO USE W/C FOR APPTS. THAT WOULD REQUIRE WALKING A LONG DISTANCE. | | |
| 06/18 | —P.T. CONSULT SENT TO ALLOW PT. TO CONTINUE PROGRAM SHE FOLLOWS AT HOME, PT. HAS APPT. THERE QD AT 9AM. | | |
| 06/18 | —TALK WITH PT. ABOUT OBTAINING A W/C AT HOME TO INCREASE HER MOBILITY. SUGGEST SHE SPEAK WITH SOCIAL WORKER ABOUT HOW TO OBTAIN ONE IN HER AREA. | | |

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06/18 —PROVIDE PT. WITH ARTHRITIS TEACHING AID THAT DEMONSTRATES NUMEROUS POSITIONS FOR INTERCOURSE. REVIEW THIS INFO. WITH HER.

**06/18 LACK OF KNOWLEDGE OF DIAGNOSTIC TESTS**

| EXPECTED OUTCOMES | C/P: | D/L: |
|---|---|---|
| 06/18 STATES WHAT TO EXPECT PRE/POST TEST | QTEST | 6/25 |
| 06/18 —PT. WILL REQUEST INFORMATION R/T HIP REPLACEMENT PROCEDURE. | TH | 6/21 |

NURSING ACTIONS

06/18 INFORM PT OF TESTS AS SOON AS POSSIBLE
06/18 ALLOW TIME FOR EXPLANATION OF TESTS
06/18 ALLOW TIME FOR ASKING QUESTIONS
06/18 PROVIDE LITERATURE WHEN APPROPRIATE
06/18 —ENCOURAGE PT. TO SPEAK WITH MD ABOUT THE HIP REPLACEMENT—PROCEDURE, HOW IT IS DONE, WHERE IT COULD BE DONE, RECUPERATION PERIOD, ETC.

**06/18 COPING, INEFFECTIVE INDIVIDUAL: MOOD ALTERATION**

| EXPECTED OUTCOMES | C/P: | D/L: |
|---|---|---|
| 06/18 V-FEELINGS OF MOOD ALTERATION DEPRESSION | QD | 6/25 |
| 06/18 V-AID(S) IN COPING WITH MOOD ALTERATION | QOD | 6/25 |

NURSING ACTIONS

06/18 ENCOURAGE EXPRESSION/DISCUSSION OF FEELINGS
06/18 PROMOTE APPROPRIATE ASSOCIATION WITH OTHERS
06/18 HELP PT TO IDENTIFY STRENGTHS & RESOURCES
06/18 AID IN PLANNING ACTIVITIES
06/18 ENCOURAGE PARTICIPATION IN DIVERSIONAL ACTIVITIES
06/18 —EXPLORE FEELING REGARDING PROFESSIONAL HELP AT HOME, SUGGEST THAT SOC. WORKER ALSO EXPLORE THIS WITH PT., ASK HER TO EXPORE PAYMENT/INSURANCE HELP.

POTENTIAL PROBLEMS & MISC PT DATA:

06/18 POTENTIAL PROBLEMS—PT. HAS HISTORY OF THROMBOCYTOPENIA, LEUKOPENIA, AND ANEMIA. ALL FEVERS MUST BE REPORTED, OBSERVE FOR UNUSUAL BLEEDING.
06/18 MISC PT DATA—PT. WILL RECEIVE FIVE SMALL MEALS A DAY—DIETARY HAS ALREADY BEEN NOTIFIED AND HAS ASSESSED PT.

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**Name:** Patient B Appendix 3

**Diagnosis:** Gaucher's disease Type 1

## I. General Information

1. Admission vital signs:
   Temperature: 36.8
   Pulse: 96
   Respirations: 28
   Blood Pressure: 98/50
2. Reason for admission: Yearly diagnostic testing
3. Medications: penicillin VK 250 mg bid
4. Allergies: None

## II. Assessment

### A. Air

1. Respiratory problems? Respirations are distinctly audible. Average 22-28/minute, SOB on exertion, no cyanosis/clubbing.

### B. Circulation

1. History gross hepatomegaly, has had three episodes in last 4 months of profuse bleeding from esophageal varices, sclerosing procedure done two times, each time patient vomited blood as first symptom
2. Aids? Normal CBC, elevated FTS's. Poor veins!! Recent difficulties. Cross-matching blood secondary to antibodies

### C. Food/Fluid

1. Eating problems? Due to early satiety eats 6 small meals/day
2. Problems with food preparation? Mother prepares meals
3. Appetite? Good
4. Nausea/vomiting? Not usually - recent vomiting was secondary to the bleeding
5. Weight change? None
6. Special diet? No
7. Aids? Dietician to see pt. regarding preferences/small meals

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**D. Elimination (Bowel/Bladder)**

1. *Bladder*
   a. Incontinence/retention? Occ. incontinent at night, uses a toddler size pamper
   b. Aids? Diaper for bedtime
2. *Bowel*
   a. Pattern (last stool?) qd - no diarrhea
   b. Blood in stool? Had black stools at time of GI bleed

**E. Neuro/Sensory**

1. Increased strength/weakness of any muscle group? Normal strength according to mother. Could not test right leg or left arm.
2. Level of consciousness and orientation? A and O x 3
3. Verbal and written comprehension? Can't read
4. Speech problems? No
5. Involuntary movements/rigidity? None
6. Type of seizures? None
7. Loss of sensation to heat, cold, touch or position? Intact
8. Visual changes? Question in past of oculomotor disturbance, EOM's intact today, no nystagmus
9. Hearing changes? No
10. Taste changes? No
11. Family history of neurological disease? No

**F. Sleep/Rest**

1. Problems? None - mother will take him on pass each evening to stay with her overnight

**G. Physical Safety**

1. Problems? Predisposed to fractures secondary to bone changes

**H. Mobility**

1. Limitations? Currently just starting to bear weight on his R foot, will walk very short distance, uses stroller, mother lifts him for transfers
2. Aids? Stroller

**I. Comfort/Pain**

1. Pain description? Has pain in R foot and L shoulder - mother thought these were bone crises as patient has a history of these, but x-rays revealed fractures. Currently on no analgesia

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**J. Hygiene/Skin**

1. ADL management? Sits in tub to wash, needs lifted in and out, he will assist with dressing, can brush own teeth. Mother will assist with ADL needs
2. Skin integrity? Intact
3. Oral mucosa? Intact
4. Ecchymotic areas? None

**K. Sexuality**

1. Genetic counseling? Mother/father aware of genetic concerns with the disease

**L. Socio/Psychological**

1. Emotional status? Very shy, frequently will not talk to strangers, becomes anxious about venipunctures
2. How are you managing with your illness? Have LMD's to handle normal care and to handle recent problems with GI bleeding. Have local rescue unit to transport patient to emergency care when needed
3. What concerns you most about your disease? Mother concerned about Matt's recent bleeds, problem with cross-matching blood
4. Who is most supportive? Mother here with her mother, receives support from family, church
5. How does your illness affect your occupation? Has been attending kindergarten

**M. Teaching/Learning**

1. Knowledge and understanding of disease? Could use more information regarding cause of varices and bleeding
2. Knowledge and understanding of medications? Understands need for penicillin
3. Knowledge and understanding of diet? Understands why he tolerates small meals better
4. Knowledge and understanding of safety measures? Aware of need to handle limbs carefully
5. What do you see as your most important learning need? Information regarding results of diagnostic tests

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## NURSING CARE PLAN

Appendix 4

**REAL & POTENTIAL PROBLEMS:**

**06/18 TIP #1—POTENTIAL FOR PATHOLOGICAL FRACTURES SECONDARY TO GAUCHER'S DISEASE.**

| | | C/P: | D/L: |
|---|---|---|---|
| EXPECTED OUTCOMES | | | |
| 06/18 | —NO PHYSICAL INJURY | QOD | 6/23 |
| NURSING ACTIONS | | | |
| 06/18 | —ALLOW FAMILY TO DO PT. TRANSFERS IF HE NEEDS TO BE PICKED UP. | | |
| 06/18 | —USE STROLLER FOR APPTS. | | |
| 06/18 | —PT. HAS HAD RECENT FX. OF LEFT SHOULDER AND RIGHT FOOT—PLEASE BE ALERT TO COMPLAINTS OF PAIN OR CHANGES IN THESE AREAS. | | |
| 06/18 | —PT. IS BEGINNING TO BEAR WT. ON HIS FOOT—ALLOW HIM TO AMBULATE AS TOLERATED. | | |
| 06/18 | —PLEASE HANDLE PT. CAREFULLY IF NURSING STAFF DO TRANSFERS. | | |

**06/18 TIP #2—POTENTIAL OF BLEEDING FROM ESOPHAGEAL VARICES.**

| | | C/P: | D/L: |
|---|---|---|---|
| EXPECTED OUTCOMES | | | |
| 06/18 | —SIGNS OF GI BLEEDING WILL BE REPORTED IMMEDIATELY. | QOD | 6/24 |
| NURSING ACTIONS | | | |
| 06/18 | —HEMATEST ALL EMESIS. | | |
| 06/18 | —NOTIFY MD. IMMEDIATELY IF IT IS POSITIVE. | | |
| 06/18 | —PLACE PT. ON BEDREST AND MONITOR VS IF THIS OCCURS. ALSO INITIATE DRAWING OF TYPE AND CROSS FOR BLOOD TRANSFUSIONS THAT MAY BE NEEDED. | | |

**06/18 LACK OF KNOWLEDGE OF—DIAGNOSTIC TESTS.**

| | | C/P: | D/L: |
|---|---|---|---|
| EXPECTED OUTCOMES | | | |
| 06/18 | —STATES WHAT TO EXPECT PRE—POST TEST—INFORM PT. AND MOTHER OF TESTS AS SOON AS POSSIBLE. | QTEST | 6/24 |

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NURSING ACTIONS

06/18 —ALLOW TIME FOR EXPLANATION OF TESTS. ALLOW TIME FOR ASKING QUESTIONS.

06/18 —PROVIDE LITERATURE WHEN APPROPRIATE—ENCOURAGE MOTHER TO SPEAK WITH MD ABOUT TEST RESULTS

06/18 —PT. HAS POOR VENOUS ACCESS, PLEASE CONSOLIDATE TESTS THAT—REQUIRE VENIPUNCTURE WHEN SCHEDULING IS DONE.

POTENTIAL PROBLEMS & MISC PT DATA:

06/18 POTENTIAL PROBLEMS—HAS BLED PROFUSELY IN PERIOD OF LAST FOUR MONTHS SECONDARY TO ESOPHAGEAL VARICES. HAVE ALREADY COMPLETED TEACHING ON CAUSE OF VARICES AND THE REASON THEY CAN HEMORRHAGE.

06/18 MISC PT DATA—PT'S MOTHER WILL RETURN PT. FROM PASS QAM FOR HIS FIRST APPT., MAY ARRIVE BEFORE VISITING HRS. WILL TAKE PT. OUT ON PASS QPM AFTER TESTS.

06/18 POTENTIAL PROBLEMS—PT. HAS HISTORY OF BONY CRISES. BE ALERT TO ACUTE BONE PAIN.

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## Examples of Helpful Agencies

Appendix 5

**March of Dimes Birth Defects Foundation**
1275 Mamaroneck Avenue
White Plains, New York 10605
(914) 428-7100

**The National Easter Seal Society, Inc.**
2023 West Ogden Avenue
Chicago, Illinois 60612
(312) 243-8400

**Epilepsy Foundation of America**
4351 Garden City Drive
Landover, Maryland 20785
(301) 459-3700

**National Foundation for Jewish Genetic Diseases**
250 Park Avenue
New York, New York 10177
(212) 682-5550

**National Genetics Foundation, Inc.**
Room 1240
555 West 57th Street
New York, New York 10019
(212) 586-5800
Home Health Services
County/Regional Home Health Services
Local Jewish Social Services

**Medic Alert Foundation International**
P.O. Box 1009
Turlock, California 95381-1009
(800) 344-3226
Local Medical Equipment Companies
Local Physical Therapy and Occupational Therapy Departments

**National Lipid Foundation**
1201 Corbin Street
Elizabeth, New Jersey 07201
Local Mental Health Clinics
Local Meals on Wheels Programs

**Gaucher Disease Registry Newsletter**
4418 Chapman Avenue
Suite 139
Orange, California 92669

**National Gaucher Foundation**
1424 K. Street
4th Floor
Washington, D.C. 20005

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## Partial Listing of Facilities Currently Doing Gaucher Research

Appendix 6

**John A. Barranger, MD, Ph.D.**
Associate Chief,
Developmental and Metabolic Neurology Branch, NINCDS
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**Ernest Beutler, MD**
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**Robert J. Desnick, Ph.D., MD**
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Chief, Division of Medical Genetics
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**Robert E. Lee, MD**
Prebyterian-University Hospital of Pittsburgh
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**David A. Wenger, Ph.D.**
Associate Professor of Pediatrics, and Biochemistry, Biophysics and Genetics
University of Colorado
Health Sciences Center
Denver, CO. 80262

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## Glossary of Terms

**Apraxia** - Inability to carry out purposeful movements in the absence of paralysis or other motor or sensory impairment.

**Glucocerebrosidase** - Enzyme which catalyzes the hydrolytic cleavage of glucose from glucocerebroside. Glucocerebroside + $H_2O$ glucocerebrosidase ceramide + glucose. A lipid catabolizing enzyme

**Glucocerebroside** - A compound of nitrogenous base, a long chain fatty acid and a sugar (glucose). It is abundant in membranes of nervous tissue, especially the myelin sheath, and is a major component of the cell membranes.

**Dysphagia** - Difficulty in swallowing.

**Dysphasia** - Impairment of ability to understand and use the symbols of language, both spoken and written.

**Fibroblast** - A connective tissue cell frequently grown in tissue culture.

**Hypersplenism** - A condition characterized by exaggeration of the suggested inhibitory or destructive functions of the spleen, resulting in deficiency of the peripheral blood elements, singly or in combination, hypercellularity of the bone marrow, and usually, but not always, splenomegaly.

**Hypertonia** - Abnormally increased muscle tone.

**Lysosome** - One of the minute bodies seen within cells, containing various hydrolytic enzymes and normally involved in the process of localized intracellular digestion.

**Lysosomal enzymes** -Enzymes found in the lysosomes.

**Macrophage** - Any of the large, highly phagocytic cells with a small, oval, sometimes indented nucleus and inconspicuous nucleoli, occurring in the walls of the blood vessels and in loose connective tissue. They are components of the reticuloendothelial system and usually immobile, but when stimulated by inflammation become actively mobile.

**Monoclonal antibody** - Derived from a single cell, pertaining to a single clone. An immunoglobulin molecule that has a specific amino acid sequence by virtue of which it interacts only with the antigen that induced its synthesis in lymphoid tissue, or with antigen closely related to it. Antibodies are classified according to their mode of action.

**Oculomotor apraxia** - Apraxia pertaining to eye movements.

**Organelle** - A minute structure serving a specific function in the life processes of a cell.

**Pathologic reflexes** -A reflex indicative of a lesion/problem in a particular reflex arc. Example, the Babinski's reflex is dorsiflexion of the big toe on stimulating the sole of the foot; it occurs in lesions of the pyrimidal tract.

**Phagocytosis** - The uptake or envelopment of solid particles by living cells.

**Pingueculae** - A small yellowish spot on the cornea, seen usually in the elderly.

**Ptosis** - Drooping of the upper eyelid from paralysis of the third nerve or from sympathetic innervation.

**Reticuloendothelial system** - A system of cells and tissues found throughout the body, especially in the blood, general connective tissue, spleen, liver, lungs, bone marrow and lymph nodes. These cells are concerned in blood cell formation and destruction, storage of fatty materials and metabolism of iron and pigment, and play a role in inflammation and immunity. Some of the cells are motile—that is, capable of spontaneous motion—and phagocytic—they can ingest and destroy unwanted foreign material.

**Retroflexion** - The bending of an organ so that its top is thrust backward.

**Serum nontartrate - inhibitable acid phosphates** - A phosphatase that is active in an acid environment. A blood test done on serum to measure the acid phosphatase level that is not inhibited by tartrates.

**Sphingolipid** - Sphingolipids are combinations of different compounds with the hydroxyl group of ceramides, e.g., sphingomyelins (with phosphoryl choline), gangliosides (with branched-chain oligosaccharides), and cerebrosides (with glucose or galactose). They occur in membranes and in particularly high concentrations in brain and nervous tissue.

**Strabismus** - Deviation of the eye that the patient cannot overcome. Also called crossed eyes.

**Trismus** -Motor disturbance of the trigeminal nerve with difficulty in opening the mouth.

**Virchow-Robin space** - Potential spaces surrounding blood vessels for a short distance as they enter the brain.

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